OM protein - protein search, using sw model

Run on: July 16, 2003, 13:53:06 ; Search time 69 Seconds
(without alignments)
42.486 Million cell updates/sec

Title: US-09-914-213-2
Perfect score: 116
Sequence: 1 GLEISEEINEEDLKECFFDDME 22

Scoring table: BLOSUM62
Gapop 10.0 , Gapext 0.5

Searched: 908470 seqs, 133250620 residues

Total number of hits satisfying chosen parameters: 908470

Minimum DB seq length: 0
Maximum DB seq length: 2000000000

Post-processing: Minimum Match 0%
Maximum Match 100%
Listing first 45 summaries

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Pred. No. is the number of results predicted by chance to have a score greater than or equal to the score of the result being printed, and is derived by analysis of the total score distribution.

SUMMARIES

Result No.	Score	% Query Match	Length	DB	ID	Description
1	116	100.0	22	21	AAB22023	Human CFTR peptide
2	116	100.0	1480	22	AAB31723	A human cystic fib
3	116	100.0	1614	22	ABG17640	Novel human diagno
4	116	100.0	1843	22	ABG14961	Novel human diagno
5	110	94.8	1091	12	AAR13303	CFTR Y1092X. Homo
6	110	94.8	1190	12	AAR13308	CFTR 3659 del C.
7	110	94.8	1476	20	AAY33968	CFTR protein seque
8	110	94.8	1479	12	AAR11602	Mutant cystic fibr
9	110	94.8	1479	12	AAR13231	CFTR delta I507.
10	110	94.8	1479	20	AAY02279	DeltaF508 cystic f
11	110	94.8	1479	23	AAU74516	Human delta-F508-C
12	110	94.8	1480	12	AAR11115	Cystic fibrosis tr
13	110	94.8	1480	12	AAR13232	CFTR G85E. Homo s
14	110	94.8	1480	12	AAR13233	CFTR I148T. Homo
15	110	94.8	1480	12	AAR13234	CFTR G178R. Homo
16	110	94.8	1480	12	AAR13235	CFTR A455E. Homo
17	110	94.8	1480	12	AAR13297	CFTR S549R. Homo
18	110	94.8	1480	12	AAR13298	CFTR G551D. Homo
19	110	94.8	1480	12	AAR13299	CFTR R560T. Homo
20	110	94.8	1480	12	AAR13300	CFTR Y563N. Homo
21	110	94.8	1480	12	AAR13301	CFTR P574H. Homo
22	110	94.8	1480	12	AAR13302	CFTR L1077P. Homo
23	110	94.8	1480	12	AAR13894	Cystic fibrosis tr
24	110	94.8	1480	13	AAR22492	Cystic Fibrosis tr
25	110	94.8	1480	13	AAR23074	Cystic fibrosis ge
26	110	94.8	1480	15	AAR79011	CFTR encoded by Ad
27	110	94.8	1480	16	AAR72852	CFTCR. Homo sapie
28	110	94.8	1480	16	AAR72384	Cystic fibrosis tr
29	110	94.8	1480	16	AAR71095	Antibody binding a
30	110	94.8	1480	20	AAY49869	Human cystic fibro
31	110	94.8	1480	20	AAY02278	A cystic fibrosis
32	110	94.8	1480	20	AAY02280	G551D cystic fibro
33	110	94.8	1480	20	AAW97573	Cystic fibrosis tr
34	110	94.8	1480	20	AAW68012	Human cystic fibro
35	110	94.8	1480	22	AAB85431	Human CFTR amino a
36	110	94.8	1480	22	AAB68049	A cystic fibrosis
37	110	94.8	1480	23	AAU74515	Human cystic fibro
38	110	94.8	1480	23	AAU74517	Human G551D-CFTR m
39	100	86.2	836	16	AAR79835	Truncated cystic f
40	94	81.0	18	21	AAB22022	Human CFTR peptide
41	68	58.6	154	22	AAM34311	Peptide #8348 enco
42	68	58.6	154	23	ABG44143	Human peptide enco
43	54	46.6	626	22	AAU07863	Polypeptide sequen
44	51	44.0	705	22	AAU33838	Staphylococcus aur
45	51	44.0	713	22	AAU36729	Staphylococcus aur

ALIGNMENTS

RESULT 1
AAB22023
ID AAB22023 standard; peptide; 22 AA.
XX
AC AAB22023;
XX
DT 08-JAN-2001 (first entry)
XX
DE Human CFTR peptide fragment NEG2.
XX
KW Human; cystic fibrosis transmembrane conductance regulator; CFTR; CF;
KW chloride channel.
XX
OS Homo sapiens.
XX
PN WO200050591-A1.
XX
PD 31-AUG-2000.
XX
PF 24-FEB-2000; 2000WO-US04642.
XX
PR 24-FEB-1999; 99US-0121495.
XX
PA (UYCA-) UNIV CASE WESTERN RESERVE.
XX
PI Adams LM, Davis PB, Ma J;
XX
DR WPI; 2000-572090/53.
XX
PT Polypeptide useful for enhancing the open probability of cystic
PT fibrosis transmembrane conductance regulator chloride channel in cystic
PT fibrosis patients having minimally active mutant protein -
XX

PS Claim 2; Fig 2; 35pp; English.
XX
CC Defects in the cystic fibrosis transmembrane conductance regulator
CC (CFTR), are associated with cystic fibrosis (CF). CFTR is a chloride
CC channel located in the apical membrane of epithelial cells. The present
CC peptide is a fragment of human CFTR. This peptide is useful for
CC activating and opening a CFTR protein by the formation of cAMP regulated
CC chloride channel. This peptide can therefore be used as therapy for CF.
XX
SQ Sequence 22 AA;

Query Match 100.0%; Score 116; DB 21; Length 22;
Best Local Similarity 100.0%; Pred. No. 2e-09;
Matches 22; Conservative 0; Mismatches 0; Indels 0; Gaps 0;

Qy 1 GLEISEEINEEDLKECFFDDME 22
||||||||||||||||||||||
Db 1 GLEISEEINEEDLKECFFDDME 22

RESULT 2
AAB31723
ID AAB31723 standard; Protein; 1480 AA.
XX
AC AAB31723;
XX
DT 30-APR-2001 (first entry)
XX
DE A human cystic fibrosis transmembrane conductance regulator.
XX
KW Arginine-framed tripeptide; AFT; endoplasmic reticulum retention;
KW cystic fibrosis transmembrane conductance regulator; CFTR; hormone;
KW cystic fibrosis; macular dystrophy; stargardt's disease; growth factor;
KW export-incompetent protein; adenine-nucleotide binding cassette protein;
KW ABC protein; immune regulator; adhesion protein; clotting factor;
KW hemostatic regulator; intracellular transport.
XX
OS Homo sapiens.
XX
PN WO200103722-A1.
XX
PD 18-JAN-2001.
XX
PF 07-JUL-2000; 2000WO-US40324.
XX
PR 09-JUL-1999; 99US-0143090.
PR 21-JUL-1999; 99US-0146097.
XX
PA (MAYO-) MAYO FOUND MEDICAL EDUCATION & RES.
XX
PI Riordan JR, Chang X;
XX
DR WPI; 2001-123141/13.
XX
PT Novel cystic fibrosis transmembrane conductance regulator proteins,
PT useful for treating cystic fibrosis have one arginine, in an arginine
PT tripeptide sequence motif, substituted by other amino acid -
XX
PS Disclosure; Fig 6; 56pp; English.
XX
CC The present sequence represents a human cystic fibrosis transmembrane
CC conductance regulator (CFTR). Arginine-framed tripeptide (AFT) sequences
CC of CFTR contribute to endoplasmic reticulum (ER) retention or delay in
CC maturation of proteins. The AFT sequences are useful as competitive
CC inhibitors of ER retention. The peptides are useful for treating cystic
CC fibrosis. They are useful for treating a subject having or suspected of
CC having a physiological disorder (e.g. macular dystrophy and stargardt's
CC disease) associated with an export-incompetent protein such as
CC adenine-nucleotide binding cassette (ABC) protein, growth factor, immune
CC regulator, adhesion protein, hormone, clotting factor, hemostatic
CC regulator and their receptors. The AFT peptides are also useful for
CC inducing or increasing intracellular transport of an export-incompetent
CC protein in a cell e.g. a cell surface or secreted protein and preferably
CC export-incompetent CFTR. They are also useful for inhibiting degradation
CC of a secreted or cell surface protein in a cell and for detecting the
CC presence of an export-incompetent protein in a cell.
XX
SQ Sequence 1480 AA;

Query Match 100.0%; Score 116; DB 22; Length 1480;
Best Local Similarity 100.0%; Pred. No. 1.7e-07;
Matches 22; Conservative 0; Mismatches 0; Indels 0; Gaps 0;

Qy 1 GLEISEEINEEDLKECFFDDME 22
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Db 817 GLEISEEINEEDLKECFFDDME 838

RESULT 3
ABG17640
ID ABG17640 standard; Protein; 1614 AA.
XX
AC ABG17640;
XX
DT 18-FEB-2002 (first entry)
XX
DE Novel human diagnostic protein #17631.
XX
KW Human; chromosome mapping; gene mapping; gene therapy; forensic;
KW food supplement; medical imaging; diagnostic; genetic disorder.
XX
OS Homo sapiens.
XX
PN WO200175067-A2.
XX
PD 11-OCT-2001.
XX
PF 30-MAR-2001; 2001WO-US08631.
XX
PR 31-MAR-2000; 2000US-0540217.
PR 23-AUG-2000; 2000US-0649167.
XX
PA (HYSE-) HYSEQ INC.
XX
PI Drmanac RT, Liu C, Tang YT;
XX
DR WPI; 2001-639362/73.
DR N-PSDB; AAS81827.
XX
PT New isolated polynucleotide and encoded polypeptides, useful in
PT diagnostics, forensics, gene mapping, identification of mutations
PT responsible for genetic disorders or other traits and to assess
PT biodiversity -
XX
PS Claim 20; SEQ ID No 47999; 103pp; English.
XX
CC The invention relates to isolated polynucleotide (I) and
CC polypeptide (II) sequences. (I) is useful as hybridisation probes,
CC polymerase chain reaction (PCR) primers, oligomers, and for chromosome
CC and gene mapping, and in recombinant production of (II). The
CC polynucleotides are also used in diagnostics as expressed sequence tags
CC for identifying expressed genes. (I) is useful in gene therapy techniques
CC to restore normal activity of (II) or to treat disease states involving
CC (II). (II) is useful for generating antibodies against it, detecting or
CC quantitating a polypeptide in tissue, as molecular weight markers and as
CC a food supplement. (II) and its binding partners are useful in medical
CC imaging of sites expressing (II). (I) and (II) are useful for treating
CC disorders involving aberrant protein expression or biological activity.
CC The polypeptide and polynucleotide sequences have applications in
CC diagnostics, forensics, gene mapping, identification of mutations
CC responsible for genetic disorders or other traits to assess biodiversity
CC and to produce other types of data and products dependent on DNA and
CC amino acid sequences. ABG00010-ABG30377 represent novel human
CC diagnostic amino acid sequences of the invention.
CC Note: The sequence data for this patent did not appear in the printed
CC specification, but was obtained in electronic format directly from WIPO

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CC   at ftp.wipo.int/pub/published_pct_sequences.
XX
SQ   Sequence   1614 AA;

  Query Match             100.0%;  Score 116;  DB 22;  Length 1614;
  Best Local Similarity   100.0%;  Pred. No. 1.8e-07;
  Matches   22;  Conservative    0;  Mismatches    0;  Indels    0;  Gaps    0;

Qy          1 GLEISEEINEEDLKECFFDDME 22
              ||||||||||||||||||||||
Db       1531 GLEISEEINEEDLKECFFDDME 1552


RESULT 4
ABG14961
ID   ABG14961 standard; Protein; 1843 AA.
XX
AC   ABG14961;
XX
DT   18-FEB-2002  (first entry)
XX
DE   Novel human diagnostic protein #14952.
XX
KW   Human; chromosome mapping; gene mapping; gene therapy; forensic;
KW   food supplement; medical imaging; diagnostic; genetic disorder.
XX
OS   Homo sapiens.
XX
PN   WO200175067-A2.
XX
PD   11-OCT-2001.
XX
PF   30-MAR-2001; 2001WO-US08631.
XX
PR   31-MAR-2000; 2000US-0540217.
PR   23-AUG-2000; 2000US-0649167.
XX
PA   (HYSE-) HYSEQ INC.
XX
PI   Drmanac RT,  Liu C,  Tang YT;
XX
DR   WPI; 2001-639362/73.
DR   N-PSDB; AAS79148.
XX
PT   New isolated polynucleotide and encoded polypeptides, useful in
PT   diagnostics, forensics, gene mapping, identification of mutations
PT   responsible for genetic disorders or other traits and to assess
PT   biodiversity -
XX
PS   Claim 20; SEQ ID No 45320; 103pp; English.
XX
CC   The invention relates to isolated polynucleotide (I) and
CC   polypeptide (II) sequences. (I) is useful as hybridisation probes,
CC   polymerase chain reaction (PCR) primers, oligomers, and for chromosome
CC   and gene mapping, and in recombinant production of (II). The
CC   polynucleotides are also used in diagnostics as expressed sequence tags
CC   for identifying expressed genes. (I) is useful in gene therapy techniques
CC   to restore normal activity of (II) or to treat disease states involving
CC   (II). (II) is useful for generating antibodies against it, detecting or
CC   quantitating a polypeptide in tissue, as molecular weight markers and as
CC   a food supplement. (II) and its binding partners are useful in medical
CC   imaging of sites expressing (II). (I) and (II) are useful for treating
CC   disorders involving aberrant protein expression or biological activity.
CC   The polypeptide and polynucleotide sequences have applications in
CC   diagnostics, forensics, gene mapping, identification of mutations
CC   responsible for genetic disorders or other traits to assess biodiversity
CC   and to produce other types of data and products dependent on DNA and
CC   amino acid sequences. ABG00010-ABG30377 represent novel human
CC   diagnostic amino acid sequences of the invention.
CC   Note: The sequence data for this patent did not appear in the printed
CC   specification, but was obtained in electronic format directly from WIPO
CC   at ftp.wipo.int/pub/published_pct_sequences.
XX
SQ   Sequence   1843 AA;

  Query Match             100.0%;  Score 116;  DB 22;  Length 1843;
  Best Local Similarity   100.0%;  Pred. No. 2.1e-07;
  Matches   22;  Conservative    0;  Mismatches    0;  Indels    0;  Gaps    0;

Qy          1 GLEISEEINEEDLKECFFDDME 22
              ||||||||||||||||||||||
Db       1777 GLEISEEINEEDLKECFFDDME 1798


RESULT 5
AAR13303
ID   AAR13303 standard; Protein; 1091 AA.
XX
AC   AAR13303;
XX
DT   14-OCT-1991  (first entry)
XX
DE   CFTR Y1092X.
XX
KW   Deletion; mutant; diagnosis; antibodies; drug therapy.
XX
OS   Homo sapiens.
XX
FH   Key             Location/Qualifiers
FT   Domain          81..102
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PN   WO9110734-A.
XX
PD   25-JUL-1991.
XX
PF   11-JAN-1991;   91WO-CA00009.
XX
PR   10-JUL-1990;   90CA-2020817.
PR   12-JAN-1990;   90CA-2007699.
PR   01-MAR-1990;   90CA-2011253.
XX
PA   (HSCR-) HSC RES DEV CORP.
XX
PI   Tsui LC,  Rommens JM,  Kerem B;
XX
DR   WPI; 1991-238022/32.
DR   N-PSDB; AAQ13066.
XX
PT   Mutant cystic fibrosis trans-membrane conductance regulator gene
PT   - used for producing prods. for diagnosis, screening and therapy
PT   of cystic fibrosis
XX
PS   Claim 20; Page 124; 178pp; English.
XX
CC   In the LY1092X mutation a C to A change is detected at nucleotide
CC   position 3408.  This would result in protein synthesis termination
CC   at amino position 1092.  Hence the amino acid Tyr is not present
CC   in the truncated polypeptide.
CC   The mutant CF gene when expressed in cells of the human body, is
CC   associated with altered cell function which correlates with the
CC   genetic disease cystic fibrosis.
CC   See also AAQ13053-72.
XX
SQ   Sequence   1091 AA;

  Query Match             94.8%;  Score 110;  DB 12;  Length 1091;
  Best Local Similarity   95.5%;  Pred. No. 8.2e-07;
  Matches   21;  Conservative    0;  Mismatches    1;  Indels    0;  Gaps    0;

Qy          1 GLEISEEINEEDLKECFFDDME 22
              |||||||||||||||| |||||
Db        817 GLEISEEINEEDLKECLFDDME 838


RESULT 6
AAR13308
ID   AAR13308 standard; Protein; 1190 AA.
XX
AC   AAR13308;
XX
DT   14-OCT-1991  (first entry)
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XX
DE CFTR 3659 del C.
XX
KW Deletion; mutant; diagnosis; antibodies; drug therapy.
XX
OS Homo sapiens.
XX
FH Key Location/Qualifiers
FT Domain 81..102
FT /label= membrane-spanning_domain
FT Domain 118..138
FT /label= membrane-spanning_domain
FT Domain 195..215
FT /label= membrane-spanning_domain
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FT /note= "by protein kinases C"
FT Modified-site 791
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FT /label= phosphorylation_site
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FT /label= phosphorylation_site
FT /note= "by protein kinases C"
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FT Modified-site 1095
FT /label= phosphorylation_site
FT /note= "by protein kinases C"
FT Modified-site 1178
FT /label= phosphorylation_site
FT /note= "by protein kinases C"
XX
PN WO9110734-A.
XX
PD 25-JUL-1991.
XX
PF 11-JAN-1991; 91WO-CA00009.
XX
PR 10-JUL-1990; 90CA-2020817.
PR 12-JAN-1990; 90CA-2007699.
PR 01-MAR-1990; 90CA-2011253.
XX
PA (HSCR-) HSC RES DEV CORP.
XX
PI Tsui LC, Rommens JM, Kerem B;
XX
DR WPI; 1991-238022/32.
DR N-PSDB; AAQ13072.
XX
PT Mutant cystic fibrosis trans-membrane conductance regulator gene
PT - used for producing prods. for diagnosis, screening and therapy
PT of cystic fibrosis
XX
PS Claim 20; Page 124; 178pp; English.

```
XX
CC   3659 del C is a frameshift mutation in exon 19.
CC   The 3659 del C mutation results in a shortened polypeptide
CC   significantly different from the single amino acid deletions or
CC   alterations.
CC   The mutant CF gene when expressed in cells of the human body, is
CC   associated with altered cell function which correlates with the
CC   genetic disease cystic fibrosis.
CC   See also AAQ13053-72.
XX
SQ   Sequence   1190 AA;

  Query Match             94.8%;  Score 110;  DB 12;  Length 1190;
  Best Local Similarity   95.5%;  Pred. No. 9e-07;
  Matches   21;  Conservative    0;  Mismatches    1;  Indels    0;  Gaps    0;

Qy          1 GLEISEEINEEDLKECFFDDME 22
              |||||||||||||||| |||||
Db        817 GLEISEEINEEDLKECLFDDME 838


RESULT 7
AAY33968
ID   AAY33968 standard; Protein; 1476 AA.
XX
AC   AAY33968;
XX
DT   26-MAY-2000  (first entry)
XX
DE   CFTR protein sequence.
XX
KW   AAV vector; inverted terminal repeat; ITR; gene therapy; CFTR; TK gene;
KW   cystic fibrosis transmembrane conductance regulator; cystic fibrosis;
KW   promoter; HSV; thymidine kinase; chromosome 7q31.
XX
OS   Homo sapiens.
XX
FH   Key             Location/Qualifiers
FT   Misc-difference 1150
FT                   /note= "encoded by AGC"
XX
PN   WO9943789-A1.
XX
PD   02-SEP-1999.
XX
PF   25-FEB-1999;   99WO-US04212.
XX
PR   25-FEB-1998;   98US-0075980.
XX
PA   (REGC ) UNIV CALIFORNIA.
XX
PI   Dong J,  Kan YW;
XX
DR   WPI; 1999-550866/46.
DR   N-PSDB; AAZ11643.
XX
PT   Efficient AAV vectors useful in gene therapy protocols for the
PT   treatment of cystic fibrosis -
XX
PS   Example 1; Page 33; 34pp; English.
XX
CC   The invention provides efficient AAV vectors with improved capacity for
CC   DNA due to the removal of all nucleic acid sequences that are not
CC   essential for replication (to leave just 2 inverted terminal repeat
CC   sequences (ITRs)). The AAV vectors may be used for the delivery of
CC   therapeutic nucleic acids in gene therapy protocols. In particular,
CC   they may be used to deliver cystic fibrosis (CF) transmembrane
CC   conductance regulator (CFTR) polynucleotides to the respiratory tract
CC   of CF patients to rectify mutations in the patients own CFTR genes and
CC   restore normal function to the chloride channel the gene encodes. The AAV
CC   vector lacks all nucleic acids that are not essential for replication,
CC   therefore giving it a greater capacity for exogenous DNA and hence
CC   improving the efficiency with which it transfects cells. The AAV vectors
CC   of the invention can efficiently and persistently transfer CFTR
CC   polynucleotides to the airway epithelium of CF patients without any
CC   adverse side effects. The present sequence represents the CFTR protein.
XX
SQ   Sequence   1476 AA;

  Query Match             94.8%;  Score 110;  DB 20;  Length 1476;
  Best Local Similarity   95.5%;  Pred. No. 1.1e-06;
  Matches   21;  Conservative    0;  Mismatches    1;  Indels    0;  Gaps    0;

Qy          1 GLEISEEINEEDLKECFFDDME 22
              |||||||||||||||| |||||
Db        817 GLEISEEINEEDLKECLFDDME 838


RESULT 8
AAR11602
ID   AAR11602 standard; Protein; 1479 AA.
XX
AC   AAR11602;
XX
DT   22-MAY-1991  (first entry)
XX
DE   Mutant cystic fibrosis transmembrane conductance regulatory protein.
XX
KW   Cystis fibrosis; transmembrane conductance regulatory protein; CFTR;
KW   diagnosis; mutant.
XX
OS   Homo sapiens.
XX
FH   Key             Location/Qualifiers
FT   Domain          81..102
FT                   /label= potential_membrane-spanning_segment
FT   Domain          118..138
FT                   /label= potential_membrane-spanning_segment
FT   Domain          195..215
FT                   /label= potential_membrane-spanning_segment
FT   Domain          221..241
FT                   /label= potential_membrane-spanning_segment
FT   Domain          308..328
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FT   Domain          330..350
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FT   Domain          911..931
FT                   /label= potential_membrane-spanning_segment
FT   Domain          990..1010
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FT                   /label= protein_kinase_C_phosphorylation_site
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FT   Misc-difference 808..808
FT                   /label= protein_kinase_C_phosphorylation_site
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PN   WO9102796-A.
XX
PD   07-MAR-1991.
XX
PF   20-AUG-1990;    90WO-CA00267.
XX
PR   31-AUG-1989;    89US-0401609.
PR   22-AUG-1989;    89US-0396894.
PR   24-AUG-1989;    89US-0399945.
XX
PA   (HSCR-) HSC RES DEV CORP.
PA   (UNMI ) UNIV OF MICHIGAN.
XX
PI   Tsui LC,  Riordan JR,  Collins FS,  Rommens JM,  Iannuzzi MC;
PI   Kerem BS,  Drumm ML,  Buchwald M;
XX
DR   WPI; 1991-087280/12.
DR   N-PSDB; AAQ11371.
XX
PT   Cystic fibrosis gene - used to produce prods. for screening,
PT   detection, diagnosis, therapy and studying cystic fibrosis
XX
PS   Disclosure; Fig 1; 163pp; English.
XX
CC   This sequence lacks amino acid Phe 508 of the normal protein, as a
CC   result of a 3bp deletion in the nucleotide sequence.
CC   The CF gene and its gene prod., nucleic acid probes and antibodies
CC   to the gene prod. can be used for screening and detection of CF
CC   carriers, CF diagnosis, prenatal CF screening and diagnosis,
CC   and gene and drug therapy.  The prods. can also be used to develop
CC   improved methods of treatment and to study the disease.
CC   See AAQ11046 for the normal CF gene and AAQ11047-48 for CF probes.
XX
SQ   Sequence   1479 AA;

  Query Match             94.8%;  Score 110;  DB 12;  Length 1479;
  Best Local Similarity   95.5%;  Pred. No. 1.1e-06;
  Matches   21;  Conservative    0;  Mismatches    1;  Indels    0;  Gaps    0;

Qy          1 GLEISEEINEEDLKECFFDDME 22
              |||||||||||||||| |||||
Db        816 GLEISEEINEEDLKECLFDDME 837


RESULT 9
AAR13231
ID   AAR13231 standard; Protein; 1479 AA.
XX
AC   AAR13231;
XX
DT   14-OCT-1991  (first entry)
XX
DE   CFTR delta I507.
XX
KW   Deletion; mutant; diagnosis; antibodies; drug therapy;
KW   ATP-binding domain.
XX
OS   Homo sapiens.
XX
FH   Key             Location/Qualifiers
FT   Domain          81..102
FT                   /label= membrane-spanning_domain
FT   Domain          118..138
FT                   /label= membrane-spanning_domain
FT   Domain          195..215
FT                   /label= membrane-spanning_domain
FT   Domain          221..241
FT                   /label= membrane-spanning_domain
FT   Domain          308..328
FT                   /label= membrane-spanning_domain
FT   Domain          330..350
FT                   /label= membrane-spanning_domain
FT   Domain          859..879
FT                   /label= membrane-spanning_domain
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FT                   /label= membrane-spanning_domain
FT   Domain          990..1010
FT                   /label= membrane-spanning_domain
FT   Domain          1013..1033
FT                   /label= membrane-spanning_domain
FT   Domain          1102..1122
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FT   Domain             1128..1149
FT                      /label= membrane-spanning_domain
FT   Domain             433..583
FT                      /label= ATP-binding_domain
FT   Domain             1218..1385
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FT   Modified-site      808
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FT                      /note= "by protein kinases C"
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XX
PN   WO9110734-A.
XX
PD   25-JUL-1991.
XX
PF   11-JAN-1991;   91WO-CA00009.
XX
PR   10-JUL-1990;   90CA-2020817.
PR   12-JAN-1990;   90CA-2007699.
PR   01-MAR-1990;   90CA-2011253.
XX
PA   (HSCR-) HSC RES DEV CORP.
XX
PI   Tsui LC,  Rommens JM,  Kerem B;
XX
DR   WPI; 1991-238022/32.
DR   N-PSDB; AAQ13053.
XX
PT   Mutant cystic fibrosis trans-membrane conductance regulator gene
PT   - used for producing prods. for diagnosis, screening and therapy
PT   of cystic fibrosis
XX
PS   Claim 20; Page 124; 178pp; English.
XX
CC   The deletion of the amino acid at the I506 or I507 position
```

```
CC   results in the loss of an isoleucine residue from the putative
CC   CFTR, within the same ATP-binding domain where deltaF508 resides,
CC   but it is not evident whether this deleted amino acid corresponds
CC   to the position 506 or 507.  Since the 506 and 507 positions are
CC   repeats, it is at present impossible to determine in which position
CC   the deletion occurs.
CC   The mutant CF gene when expressed in cells of the human body, is
CC   associated with altered cell function which correlates with the
CC   genetic disease cystic fibrosis.
CC   See also AAQ13053-72.
XX
SQ   Sequence   1479 AA;

  Query Match             94.8%;  Score 110;  DB 12;  Length 1479;
  Best Local Similarity   95.5%;  Pred. No. 1.1e-06;
  Matches   21;  Conservative    0;  Mismatches    1;  Indels    0;  Gaps    0;

Qy          1 GLEISEEINEEDLKECFFDDME 22
              ||||||||||||||| |||||
Db        816 GLEISEEINEEDLKECLFDDME 837


RESULT 10
AAY02279
ID   AAY02279 standard; Protein; 1479 AA.
XX
AC   AAY02279;
XX
DT   08-JUL-1999  (first entry)
XX
DE   DeltaF508 cystic fibrosis transmembrane conductance regulator.
XX
KW   Flavone; isoflavone; reservatrol; ascorbic acid; ascorbate salt;
KW   dehydroascorbic acid; chloride transport; epithelial cell;
KW   cystic fibrosis; chloride ion conductance;
KW   cystic fibrosis transmembrane conductance regulator; CFTR;
KW   chronic bronchitis; asthma; intestinal constipation.
XX
OS   Homo sapiens.
XX
PN   WO9918953-A1.
XX
PD   22-APR-1999.
XX
PF   16-OCT-1998;   98WO-US21887.
XX
PR   16-OCT-1997;   97US-0951912.
XX
PA   (CHIL-) CHILDREN'S HOSPITAL OAKLAND RES INST.
XX
PI   Fischer HB,  Illek B;
XX
DR   WPI; 1999-277427/23.
DR   N-PSDB; AAX35553.
XX
PT   Use of flavones and isoflavones - for stimulating chloride transport
PT   in epithelial cells and treating cystic fibrosis
XX
PS   Disclosure; Page 73-80; 97pp; English.
XX
CC   The specification describes compounds comprising flavones/isoflavones,
CC   reservatrol, ascorbic acid, ascorbate salts and/or dehydroascorbic
CC   acid which can be used for stimulating chloride transport in epithelial
CC   cells and treating cystic fibrosis. The compounds can be used to
CC   increase chloride ion conductance in airway epithelial cells or
CC   intestine, pancreas, gallbladder, sweat duct, salivary gland or mammary
CC   epithelial cells. The compounds are useful for treating a patient with
CC   cystic fibrosis, where the patient's cystic fibrosis transmembrane
CC   conductance regulator (CFTR) protein has a deletion at position 508 or
CC   point mutation at 551. They may also be used for treating chronic
CC   bronchitis, asthma and intestinal constipation. The present sequence
CC   represents a human CTFR protein with a F508 deletion mutation.
XX
SQ   Sequence   1479 AA;

  Query Match             94.8%;  Score 110;  DB 20;  Length 1479;
  Best Local Similarity   95.5%;  Pred. No. 1.1e-06;
  Matches   21;  Conservative    0;  Mismatches    1;  Indels    0;  Gaps    0;

Qy          1 GLEISEEINEEDLKECFFDDME 22
              ||||||||||||||| |||||
Db        816 GLEISEEINEEDLKECLFDDME 837


RESULT 11
AAU74516
ID   AAU74516 standard; Protein; 1479 AA.
XX
AC   AAU74516;
XX
DT   23-APR-2002  (first entry)
XX
DE   Human delta-F508-CFTR mutant polypeptide.
XX
KW   Human; cystic fibrosis transmembrane conductance regulator; CFTR;
KW   flavone; isoflavone; chloride transport; epithelial tissue; mucus;
KW   cystic fibrosis; chronic bronchitis; asthma; delta-F508-CFTR; protein.
XX
OS   Homo sapiens.
XX
PN   US6329422-B1.
XX
PD   11-DEC-2001.
XX
PF   16-OCT-1998;   98US-0174077.
XX
PR   16-OCT-1997;   97US-0951912.
XX
PA   (CHIL-) CHILDREN'S HOSPITAL OAKLAND RES INST.
XX
PI   Fischer H,  Illek B;
XX
DR   WPI; 2002-105224/14.
DR   N-PSDB; AAS20529.
XX
PT   Pharmaceutical composition for the treatment of cystic fibrosis
PT   comprises flavones or isoflavones -
XX
PS   Disclosure; Column 37-44; 50pp; English.
XX
CC   The invention relates to a pharmaceutical composition comprising one or
CC   more compounds such as flavones or isoflavones, capable of stimulating
CC   chloride transport in epithelial tissues, for treatment of cystic
CC   fibrosis and other diseases associated with excessive accumulation of
CC   mucus, e.g. chronic bronchitis and asthma. The active compound increases
CC   expression of a cystic fibrosis transmembrane conductance regulator
CC   (CFTR) in an epithelial cell and/or acts as a chemical chaperone that
CC   increases trafficking of a CFTR to a plasma membrane in an epithelial
CC   cell. This sequence represents the human delta-F508-CFTR mutant
CC   polypeptide of the invention.
XX
SQ   Sequence   1479 AA;

  Query Match             94.8%;  Score 110;  DB 23;  Length 1479;
  Best Local Similarity   95.5%;  Pred. No. 1.1e-06;
  Matches   21;  Conservative    0;  Mismatches    1;  Indels    0;  Gaps    0;

Qy          1 GLEISEEINEEDLKECFFDDME 22
              ||||||||||||||| |||||
Db        816 GLEISEEINEEDLKECLFDDME 837


RESULT 12
AAR11115
ID   AAR11115 standard; Protein; 1480 AA.
XX
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AC   AAR11115;
XX
DT   22-MAY-1991  (first entry)
XX
DE   Cystic fibrosis transmembrane conductance regulatory protein.
XX
KW   Cystis fibrosis; transmembrane conductance regulatory protein; CFTR;
KW   diagnosis.
XX
OS   Homo sapiens.
XX
FH   Key             Location/Qualifiers
FT   Domain          81..102
FT                   /label= potential_membrane-spanning_segment
FT   Domain          118..138
FT                   /label= potential_membrane-spanning_segment
FT   Domain          195..215
FT                   /label= potential_membrane-spanning_segment
FT   Domain          221..241
FT                   /label= potential_membrane-spanning_segment
FT   Domain          308..328
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FT   Domain          330..350
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FT   Domain          860..880
FT                   /label= potential_membrane-spanning_segment
FT   Domain          912..932
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FT   Domain          991..1011
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FT   Domain          433..584
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FT   Misc-difference 63..63
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FT   Misc-difference 168..168
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PN   WO9102796-A.
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PD   07-MAR-1991.
XX
PF   20-AUG-1990;   90WO-CA00267.
XX
PR   31-AUG-1989;   89US-0401609.
PR   22-AUG-1989;   89US-0396894.
PR   24-AUG-1989;   89US-0399945.
XX
PA   (HSCR-) HSC RES DEV CORP.
PA   (UNMI ) UNIV OF MICHIGAN.
XX
PI   Tsul LC,  Riordan JR,  Collins FS,  Rommens JM,  Iannuzzi MC;
PI   Kerem BS,  Drumm ML,  Buchwald M;
XX
DR   WPI; 1991-087280/12.
DR   P-PSDB; AAR11115.
XX
PT   Cystic fibrosis gene - used to produce prods. for screening,
PT   detection, diagnosis, therapy and studying cystic fibrosis
XX
PS   Disclosure; Fig 1; 163pp; English.
XX
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CC   The protein has a mol.wt. of 170 kD and has two repeated motifs,
CC   each comprising a set of amino acids comprising six highly
CC   hydrophobic segments (see membrane spanning segments in features),
CC   capable of spanning a lipid bylayer of an epithelial cell membrane
CC   several times followed by an amino acid sequence constituting
CC   a nucleotide (ATP)-binding fold.  (The first of the (ATP)-binding
CC   folds has the Phe deletion (See AAR11602 for the mutant protein)).
CC   Between the repeated motifs is a highly charged cytoplasmic domain.
CC   The CF gene and its gene prod., nucleic acid probes and antibodies
CC   to the gene prod. can be used for screening and detection of CF
CC   carriers, CF diagnosis, prenatal CF screening and diagnosis,
CC   and gene and drug therapy.  The prods. can also be used to develop
CC   improved methods of treatment and to study the disease.
CC   See AAQ11371 for the mutant CF gene and AAQ11047-48 for CF probes.
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SQ   Sequence   1480 AA;

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Db        817 GLEISEEINEEDLKECLFDDME 838


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AC   AAR13232;
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DT   14-OCT-1991  (first entry)
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DE   CFTR G85E.
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KW   Deletion; mutant; diagnosis; antibodies; drug therapy.
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OS   Homo sapiens.
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PR   12-JAN-1990;   90CA-2007699.
PR   01-MAR-1990;   90CA-2011253.
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PA   (HSCR-) HSC RES DEV CORP.
XX
PI   Tsui LC,  Rommens JM,  Kerem B;
XX
DR   WPI; 1991-238022/32.
DR   N-PSDB; AAQ13054.
XX
PT   Mutant cystic fibrosis trans-membrane conductance regulator gene
PT   - used for producing prods. for diagnosis, screening and therapy
PT   of cystic fibrosis
XX
PS   Claim 20; Page 124; 178pp; English.
XX
CC   The G85E mutation in exon3 involves a G to A transition at
CC   nucleotide position 386.  The predicted Gly to Glu amino acid
CC   change is associated with a group IIb haplotype.  The mutation
CC   destroys a Hinf1 site.
CC   The mutant CF gene when expressed in cells of the human body, is
CC   associated with altered cell function which correlates with the
CC   genetic disease cystic fibrosis.
CC   See also AAQ13053-72.
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SQ   Sequence   1480 AA;

  Query Match             94.8%;  Score 110;  DB 12;  Length 1480;
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Qy          1 GLEISEEINEEDLKECFFDDME 22
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Db        817 GLEISEEINEEDLKECLFDDME 838
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DT   14-OCT-1991  (first entry)
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DE   CFTR I148T.
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PF   11-JAN-1991;   91WO-CA00009.
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PR   12-JAN-1990;   90CA-2007699.
PR   01-MAR-1990;   90CA-2011253.
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XX
PI   Tsui LC,  Rommens JM,  Kerem B;
XX
DR   WPI; 1991-238022/32.
DR   N-PSDB; AAQ13055.
XX
PT   Mutant cystic fibrosis trans-membrane conductance regulator gene
PT   - used for producing prods. for diagnosis, screening and therapy
PT   of cystic fibrosis
XX
PS   Claim 20; Page 124; 178pp; English.
XX
CC   The I148T mutation in exon 4 involves a T to C transition at
CC   nucleotide position 575 resulting in an Ile to Thr change at amino
CC   acid position 148.
CC   The mutant CF gene when expressed in cells of the human body, is
CC   associated with altered cell function which correlates with the
CC   genetic disease cystic fibrosis.
CC   See also AAQ13053-72.
XX
SQ   Sequence   1480 AA;

  Query Match             94.8%;  Score 110;  DB 12;  Length 1480;
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Qy          1 GLEISEEINEEDLKECFFDDME 22
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Db        817 GLEISEEINEEDLKECLFDDME 838


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AC   AAR13234;
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DT   14-OCT-1991  (first entry)
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DE   CFTR G178R.
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KW   Deletion; mutant; diagnosis; antibodies; drug therapy.
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PI Tsui LC, Rommens JM, Kerem B;
XX
DR WPI; 1991-238022/32.
DR N-PSDB; AAQ13056.
XX
PT Mutant cystic fibrosis trans-membrane conductance regulator gene
PT - used for producing prods. for diagnosis, screening and therapy
PT of cystic fibrosis
XX
PS Claim 20; Page 124; 178pp; English.
XX
CC The G178R mutation in exon 5 involves a G to A transition at
CC nucleotide position 664 resulting in a Gly to Arg change at amino
CC acid position 178.
CC The mutant CF gene when expressed in cells of the human body, is
CC associated with altered cell function which correlates with the
CC genetic disease cystic fibrosis.
CC See also AAQ13053-72.
XX
SQ Sequence 1480 AA;

Query Match 94.8%; Score 110; DB 12; Length 1480;
Best Local Similarity 95.5%; Pred. No. 1.1e-06;
Matches 21; Conservative 0; Mismatches 1; Indels 0; Gaps 0;

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Qy          1 GLEISEEINEEDLKECFFDDME 22
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Db        817 GLEISEEINEEDLKECLFDDME 838
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